



FGD4 gene

FYVE, RhoGEF and PH domain containing 4

Normal Function

The *FGD4* gene provides instructions for making a protein called frabin. This protein plays an important role in the nervous system, where it helps regulate a type of cell signaling involved in myelin production. Myelin is a fatty substance that insulates nerve cells and promotes the rapid transmission of nerve impulses. The formation of a protective myelin sheath around nerve cells is called myelination. Frabin is particularly important in the myelination of the peripheral nervous system, which connects the brain and spinal cord to muscles and to sensory cells that detect sensations such as touch, pain, heat, and sound.

Frabin is also believed to be involved in the organization of the actin cytoskeleton, which is a network of fibers that make up the cell's structural framework. The actin cytoskeleton has several critical functions, including determining cell shape and allowing cells to move. Researchers believe that frabin plays a role in attaching the actin cytoskeleton to the cell membrane.

Health Conditions Related to Genetic Changes

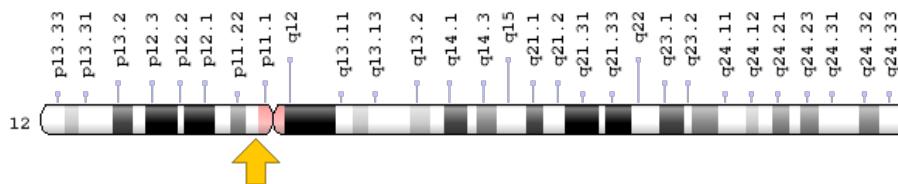
Charcot-Marie-Tooth disease

At least five mutations in the *FGD4* gene have been identified in people with Charcot-Marie-Tooth disease type 4H. These mutations change the protein's shape or result in an abnormally short, nonfunctional protein. The disruption of frabin's role in cell signaling causes abnormal myelination of the peripheral nervous system, resulting in the signs and symptoms of Charcot-Marie-Tooth disease type 4H.

Chromosomal Location

Cytogenetic Location: 12p11.21, which is the short (p) arm of chromosome 12 at position 11.21

Molecular Location: base pairs 32,399,523 to 32,646,050 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- actin-filament binding protein frabin
- CMT4H
- DKFZp313E1818
- FGD1 family, member 4
- FGD4_HUMAN
- FRABIN
- FRABP
- MGC57222
- ZFYVE6

Additional Information & Resources

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 4
<https://www.ncbi.nlm.nih.gov/books/NBK1468>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28FGD4%5BTIAB%5D%29+OR+%28CMT4H%5BTIAB%5D%29+OR+%28FRABIN%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- FYVE, RhoGEF, AND PH DOMAIN-CONTAINING PROTEIN 4
<http://omim.org/entry/611104>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_FGD4.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=FGD4%5Bgene%5D>
- HGNC Gene Family: Pleckstrin homology domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/682>
- HGNC Gene Family: Rho guanine nucleotide exchange factors
<http://www.genenames.org/cgi-bin/genefamilies/set/722>
- HGNC Gene Family: Zinc fingers FYVE-type
<http://www.genenames.org/cgi-bin/genefamilies/set/81>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19125
- Inherited Peripheral Neuropathies Mutation Database
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=41>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/121512>
- UniProt
<http://www.uniprot.org/uniprot/Q96M96>

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